



PNPLA2 gene

patatin like phospholipase domain containing 2

Normal Function

The *PNPLA2* gene provides instructions for making an enzyme called adipose triglyceride lipase (ATGL). The ATGL enzyme plays a role in breaking down fats called triglycerides, which are the main source of stored energy in cells. Triglycerides are the major component of cell structures called lipid droplets (also called adiposomes). The ATGL enzyme is found on the surface of lipid droplets. When activated, the ATGL enzyme breaks down triglycerides to provide energy for the body.

Health Conditions Related to Genetic Changes

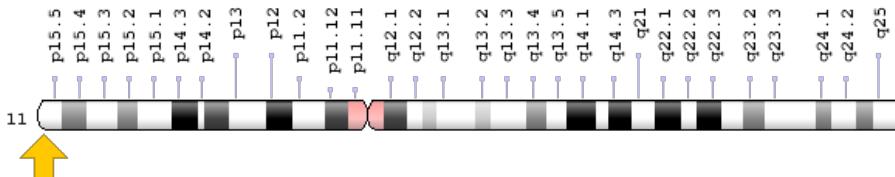
neutral lipid storage disease with myopathy

At least five mutations in the *PNPLA2* gene have been found to cause neutral lipid storage disease with myopathy. Some of these mutations cause the enzyme to function abnormally. Other mutations prevent the enzyme from ever reaching lipid droplets, so it is unable to interact with triglycerides. Any disruption in the breakdown of triglycerides leads to the accumulation of these fats in muscle and other tissues throughout the body, resulting in the signs and symptoms of neutral lipid storage disease with myopathy.

Chromosomal Location

Cytogenetic Location: 11p15.5, which is the short (p) arm of chromosome 11 at position 15.5

Molecular Location: base pairs 818,895 to 825,573 on chromosome 11 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- adipose triglyceride lipase
- ATGL
- desnutrin
- FP17548
- patatin-like phospholipase domain containing 2
- PLPL2_HUMAN
- transport-secretion protein 2.2
- triglyceride hydrolase
- TTS-2.2
- TTS2

Additional Information & Resources

Educational Resources

- Biochemistry (fifth edition, 2002): The Utilization of Fatty Acids as Fuel Requires Three Stages of Processing
<https://www.ncbi.nlm.nih.gov/books/NBK22581/>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28PNPLA2%5BTIAB%5D%29+OR+%28ATGL%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

OMIM

- PATATIN-LIKE PHOSPHOLIPASE DOMAIN-CONTAINING PROTEIN 2
<http://omim.org/entry/609059>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_PNPLA2.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=PNPLA2%5Bgene%5D>
- HGNC Gene Family: Lipases
<http://www.genenames.org/cgi-bin/genefamilies/set/464>

- HGNC Gene Family: Patatin like phospholipase domain containing
<http://www.genenames.org/cgi-bin/genefamilies/set/466>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=30802
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/57104>
- UniProt
<http://www.uniprot.org/uniprot/Q96AD5>

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